

Note: The following information is provided by the author(s) and has not been reviewed by *GeneReviews* staff.

Table 2. Normal *EMD* Allelic Variants

cDNA Change	Protein	Reference
c.(?_269)_(1090_?)inv		Small et al 1997
445 G>C	D149H	SNP / NCBI
824C>G		SNP / NCBI
825C>T		SNP / NCBI

Table 3. Normal *LMNA* Allelic Variants

cDNA Change	Protein	Reference
-128T>C	??	Benedetti 2005 (ASHG meeting 2006), base Leiden
c.5A>G	E2G	Dsnp, Leiden Muscular Dystrophy pages
51C>T	S17S	Vigouroux et al 2000, Sebillon et al 2003, Jacob et al 2005, Dsnp
75 C>T	R25R	Bonne, unpublished data
357C>T	R119R	Sebillon et al 2003
438C>T	A146A	Bonne, unpublished data
c.513+9A>G	??	GenBank, Leiden Muscular Dystrophy pages
612G>A	L204L	Eriksson et al 2003; Taylor et al 2004; Jacob et al 2005; Bonne, unpublished data
c.639+56G>A	??	Leiden Muscular Dystrophy pages
c.639+73C>T	??	Leiden Muscular Dystrophy pages
c.640-10A>C	??	Leiden Muscular Dystrophy pages
645 G>T	L215L	Bonne, unpublished data
c.691A>C	N231H	Dsnp, Leiden Muscular Dystrophy pages
c.723G>A	L241L	Eriksson et al 2003
IVS4+13G>T	??	Vigouroux et al 2000

cDNA Change	Protein	Reference
IVS4+61C>T	??	Eriksson et al 2003
IVS4-13T>A	??	Bonne, unpublished data
861T>C	A287A	Speckman et al 2000, Vigouroux et al 2000, Novelli et al 2002, Eriksson et al 2003, Jacob et al 2005
IVS5-83G>T	??	Dsnp, Leiden Muscular Dystrophy pages
1022 A>C	K341T	
1028G>A	R343Gln	Vytopil et al 2002
1098G>A	K366K	Sebillon et al 2003
IVS6+16G>A	??	Novelli et al 2002, Simha et al 2003
IVS6-43A>G	??	Dsnp, Leiden Muscular Dystrophy pages
1299C>T	H433H	Sebillon et al 2003
1338T>C	D446D	Speckman et al 2000, Vigouroux et al 2000, Novelli et al 2002, Eriksson et al 2003, Jacob et al 2005
IVS8+44C>T		Simha et al 2003
IVS8-41C>T		Vigouroux et al 2000, Eriksson et al 2003, Simha et al 2003
IVS9-55A>G		Vigouroux et al 2000
1580G>A	R527H	Novelli et al 2002
1584G>A	T528T	Bonne, unpublished data
1698C>T	H566H	Genschel & Schmidt 2000, Vigouroux et al 2000, Novelli et al 2002, Eriksson et al 2003, Shen et al 2003, Jacob et al 2005, Murase et al 2002
1761G>A	L587L	Bonne, unpublished data
1871G>T	R624L	Dsnp, Leiden Muscular Dystrophy pages
1908C>T	S636S	Hegele et al 2000 (Physiol genomics)
2074G>C		Taylor 2004, Dsnp, Leiden Muscular Dystrophy pages, present in GenBank (dbEST)
c.2135T>G		GenBank L12401, BM821790
c.2142T>A		Dsnp, Leiden Muscular Dystrophy pages
c.2187A>C		GenBank (dbEST)
c.2336G>T		Dsnp, Leiden Muscular Dystrophy pages

cDNA Change	Protein	Reference
c.2734G>A		Dsnp, Leiden Muscular Dystrophy pages
c.2910C>T		Dsnp, Leiden Muscular Dystrophy pages
c.3019C>T		Dsnp, Leiden Muscular Dystrophy pages